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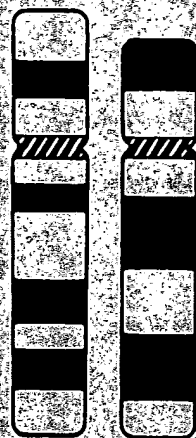
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CHROMOSOMAL VARIATION IN MAN



*A Catalog of Chromosomal
Variants and Anomalies*

7th Edition

Digamber S. Borgaonkar

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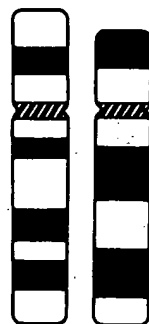
*A Catalog of Chromosomal
Variants and Anomalies*

7th Edition

CHROMOSOMAL VARIATION IN MAN

*A Catalog of Chromosomal
Variants and Anomalies*

7th Edition



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Carroll, A J, J T Prchal and W H Finley: Transient hematologic disturbance in three members of a family segregating a balanced chromosome translocation. *AJHG* 36:265(Abstract), 1984; Yang-Feng, T L, G Bruns, A J Carroll, K O J Simola and U Francke: Localization of the LDHA gene to 11p14 to 11p15 by in situ hybridization of an LDHA cDNA probe to two translocations with breakpoints in 11p13. *Hum. Genet.* 74:331-334, 1986.

Chitayat, D, C L Fagerstrom, D K Kalousek, J Rootman, G P Taylor and J G Hall: De novo reciprocal 1p;2q translocation in a child with multiple congenital anomalies/mental retardation syndrome. *AJMG* 32:36-41, 1989.
Patinet B.T.
46,XY,t(1;2)(p22;q22).
Some of the features were growth failure, mental retardation, microcephaly, cryptorchidism, partial syndactyly of the second and third toes and an unusual facial appearance.
MIM#270400

de Michelena, M I, J Villacorta and J Chavez: Double chromosome anomaly: interstitial deletion 5q and reciprocal translocation (1;11)(p22;q21). *AJMG* 36:29-32, 1990.
46,XX,t(1;11)del(5)(pter to q15::q31 to qter)de novo.

Deroover, J, J P Fryns, J Haegeman and H van den Berghe: Paracentric inversion in the short arm of chromosome 1. *Hum. Genet.* 49:117-121, 1979; Fryns, J P, and H van den Berghe: Paracentric inversion in man: personal experience and review of literature. *Hum. Genet.* 54:413-416, 1980. Family V.D.C.
46,XX,inv(1)(p22p36).
Patient MV, 19 years old was an anxious, autistic, severely retarded girl.
46,XY,inv(1)(p22p36).
46,XY,inv(1)(pter to p36::p22 to p36::p22 to qter).

Dhadial, R K, and M F Smith: Terminal 7p deletion and 1;7 translocation associated with craniosynostosis. *Hum. Genet.* 50:285-289, 1979.
46,XX,t(1;7)(p22;p15), del(7)(pter to p15).
Parental karyotypes were normal.
MIM#218500

Diedrich, U, I Hansmann, D Janke, O Opitz and H D Probeck: Chromosome anomalies in 136 couples with a history of recurrent abortions. *Hum. Genet.* 65:48-52, 1983.
46,XX,rcp(1;8)(p22;q22).

Francke, U, B de Martinville, L Coussens and A Ullrich: The Human gene for the beta subunit of nerve growth factor is located on the proximal short arm of chromosome 1. *Science* 222:1248-1251, 1983.

Fryns, J P, A Kleczkowska, E Kubien, P Petit and H Van den Berghe: Cytogenetic survey in couples with recurrent fetal wastage. *Hum. Genet.* 65:336-354, 1984.
46,XX,t(1;3)(p22;p27).

Fryns, J P, A Kleczkowska, E Kubien and H Van den Berghe: Cytogenetic findings in moderate and severe mental retardation. A study of an institutionalized population of 1991 patients. *Acta Paed. Scand. Suppl.* 313:1-23, 1984.
46,XY,t(1;9)(p22;p24)mat.
46,XX,inv(1)(p22p25)pat.

Fryns, J P, A Kleczkowska and H Van den Berghe: Paracentric inversions in man. *Hum. Genet.* 73:205-213, 1986. Family Nos. 2 and 3.
46,XX,inv(1)(p22p35)/47,XX,+21,inv(1)(p22p35)pat.
46,XX,inv(1)(p22q36)pat.

Fukushima, Y, Y Kuroki and T Ito: Balanced double complex translocations [46,XX,t(1p;6p;7p;3q;11p)(11q;22p;21q)] in an infant with multiple congenital anomalies. *AJMG* 25:313-317, 1986.
Patient (KCMC-90261) was 1 month old.
46,XX,t(1;6;7;11)(11;22;21)(1qter to p22::11p15 to 11pter;6qter to 6p21::1p22 to 1pter;7qter to 7p15::6p21 to 6pter;3pter to 3q27::7p15 to 7pter;3qter to 3q27::11p15 to 11q11::21q11 to 21qter;22qter to 22p11::11q11 to 11qter;21pter to 21q11::22p11 to 22pter).

Geiger, C J, F M Salzano, M S Mattevi, B Erdtmann and F J da Rocha: Chromosome Variation and genetic counseling-20 years of experience in Brazil. *Brazil. J. Genet.* 10:581-591, 1987.

Case 1789; I. M. F. She had 5 miscarriages. Karyotypes. 46,XX or XY,t(1;18

Same entry as in 0X recommendations ha

Kalousek, D K: In: G Wulf: Risks of un amniocentesis to can rearrangements: data laboratories. *AJMG* Observation No. 2. 46,XY,t(1;2)(p22;q3 Observation No. 79. 46,XX,t(1;10)(p22;q

McCorquodale, M I detection of de novo breakpoints as those case report. *Prenat. J* 46,XX,t(1;5)(p22;q2 Normal growth and c

Muro, R, M R Caba J Egozcue: A new b translocation in a ster 1985. Patient was 42 years 46,XY,t(1;22)(p22;q

Morichon-Delvallez, Paracentric inversion observations. *Ann. C* Observation 1: M. et 46,XY,inv(1)(p22p36

Same entry as in 01p

Neu, R L, K Miller G Wulf: Risks of un amniocentesis to can rearrangements: data laboratories. *AJMG* Observation No. 90. 46,XX,t(1;11)(p22;p1 Observation No. 150. 46,XY,t(1;18)(p22;q